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Case Report

# Atypical form of Mayer-Rokitansky-Küster-Hauser syndrome: A case report ☆

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ABSTRACT

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is a congenital disorder syndrome characterized by failure of the uterine and vaginal organs to develop normally. The prevalence of MRKH is estimated to occur in about 1 in 5000 of female live births. A 25-year-old female patient comes to a general obstetric and gynecological polyclinic with complaints of not being able to menstruate at all since birth. There is a history of vaginal discharge but it is neither viscous nor had an odor. On ultrasound examination, the structure of the uterus and ovaries was not found in a normal place. On follow up MRI examination showed agenesis of the uterus and proximal two-third of the vagina accompanied by abnormal positioning of both ovaries, thus supporting to atypical form of MRKHS. The patient is not given drug therapy, but she was planned to transplant the uterine organs. This case report suggests MRKH syndrome can be characterized by ectopic ovaries and a uterus that is not fully developed and can also be accompanied by agenesis of the vaginal organs. Pelvic ultrasound is the main modality chosen to be performed in patients with symptoms of primary amenorrhea. When pelvic organs cannot be visualized properly, it will be performed MRI examination. MRI examination is known to have sensitivity and specificity up to 100% in diagnosing MRKH syndrome. This case report describes a 25-year-old woman with primary amenorrhea with MRKH syndrome. MRI is a sensitive and specific examination to confirm the diagnosis.



## Background

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a very rare congenital abnormality syndrome and is found in female patients. It is characterized by the failure of the uterine and vaginal organs to develop normally. Women with this disorder have normal secondary sex characteristics (such as developing breasts and pubic hair), but do not experience menstrual cycles (primary amenorrhea) [1].

MRKH can be divided into type 1 and type 2. MRKH type 1 is also known as isolated Mullerian aplasia or Rokitansky

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sequence. This type has symptoms that vary from one individual to another. In many cases, the uterus and/or vagina are underdeveloped (aplasia), and in more severe cases there may be atresia of the upper portion of the vagina and underdeveloped uterus [2]. The fallopian tubes can also be affected by their normal function. In type 2 it is also called Mullerian duct aplasia, Renal dysplasia, and Cervical Somite anomalies or also known as the MURCS association. Typical symptoms of type 2 MRKH are developmental disorders of the kidneys to develop properly and malformations in the bones, especially in the vertebrae. Malformations of the heart and hearing loss can also occur but are very rare [3].

The prevalence of MRKH is estimated to occur around 1 in 5000 female newborns, however this figure is still underreported in various countries. In other literature, it is estimated that the prevalence is 1 in 4000-20,000 births. Recently, only 2 population-based studies have examined the prevalence of MRKH. Previous research with a cohort study design on 161 patients who were given treatment at 5 university-based hospitals in Finland in 1978-1993, the results showed that MRKH syndrome occurred in 1 out of 4961 female births [4]. Herlin et al. [5] reported a cohort study in Denmark who identified through searches of the National Danish Patient Registry from 1994 to 2015. The results showed a prevalence of 1 in 4982 live female births.

The primary clinical manifestation that is commonly found is primary amenorrhea. Sexual characters and external phenotypes that are also normal can cause a delayed diagnosis. Ultrasonography (US) is a first-line diagnostic tool for female patients with suspected MRKH in health facilities. Surgical techniques such as vaginoplasty are also commonly performed in patients with MRKH to correct abnormal sexual function. But generally, in making the diagnosis of MRKH, ultrasound is not very good at seeing in detail and in seeing the location of the ectopic ovary [6,7]. Laparoscopic technique with the help intraoperative ultrasound can help establish the diagnosis. Magnetic resonance imaging (MRI) is the only modality that is noninvasive and has good sensitivity and specificity in making the diagnosis of MRKH [8].

This case report discusses a 25-year-old female patient with primary amenorrhea caused by MRKH syndrome who was diagnosed by MRI examination.

## Case illustration

A 25-year-old female patient came to the general obstetrics and gynecology polyclinic with complaints of not being able to menstruate since birth. Other complaints such as abdominal pain, palpable lump, abdominal distention, lump in the vagina, pain after sexual intercourse, and bleeding from the birth canal were denied. There is a history of vaginal discharge but which was not thick or odorous. History of hypertension, diabetes mellitus, and pulmonary tuberculosis was denied by the patient. The patient also never underwent surgery, radiotherapy, or chemotherapy before.

The first day of the last menstrual period and menarche are unknown. The patient is unmarried and never has sexual intercourse at all. History of contraceptive use was also denied by the patient. On physical examination vital signs were found within normal limits.

The ovaries that are usually found around the hypogastric artery are not found on an ultrasound examination and on follow-up examination, there were bilateral ectopic ovaries, which were confirmed on MRI examination.

Ultrasound examination showed no uterus or ovaries (Fig. 1). On noncontrast MRI examination at 3 Tesla, a unilocular lesion, homogeneous in signal intensity, with well defined, regular margins, the largest diameter of 0.52 cm was seen in the left anterior abdominal area (Fig. 2A), which was hypointense on T1WI, hyperintense on T2WI and T2FS, and gave no area of restricted diffusion on DWI-ADC. Postcontrast scanning did not show any enhancement in the left ovary. A similar finding was seen with the largest diameter measuring 0.57 cm in the right anterior abdominal area denoting the right ovary (Fig. 2B).

The structure of the uterus, right and left fallopian tubes, and the proximal two-third of the vagina are not well visualized (Figs. 2 and 3). The results of the MRI examination showed agenesis of the uterus and proximal two-third of the vagina

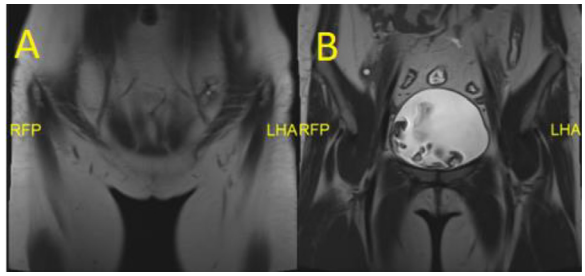


**Fig. 2 – MRI examination results in patients with MRKHS. (A) Left ectopic ovary is found. (B) There is a right ectopic ovary.**

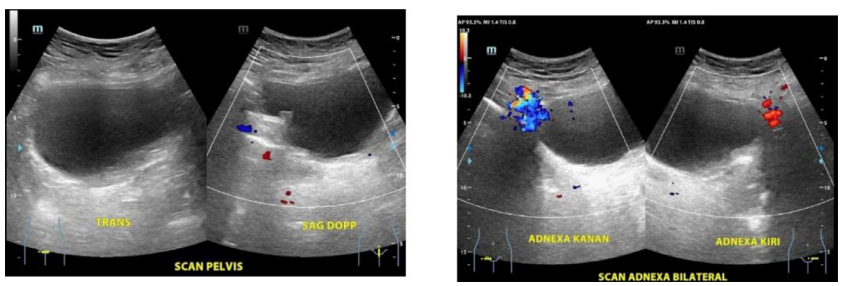


**Fig. 1 – The results of the ultrasound examination found no uterine or ovaries in the area around the hypogastric vein.**

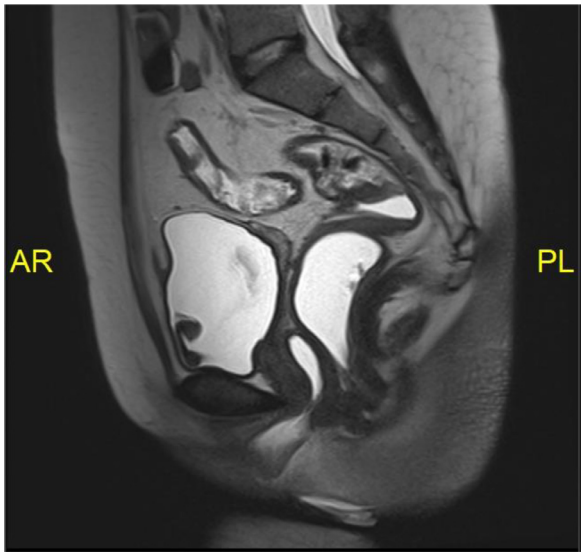


**Fig. 3 – The sagittal section of the MRI showed that the uterus was not well visualized.**

accompanied by abnormal positioning of the ovaries bilaterally, thus supporting the atypical form of MRKHS.

X-ray examination, pathological anatomy and immunohistochemistry (IHC) were not performed in this patient. The patient was not given medical therapy, but was planned to undergo uterine organ transplantation.

## Discussion

MRI examination in this study aims to confirm the diagnosis of MRKH syndrome in patients with primary amenorrhea. Ultrasound examination is generally inconclusive when compared with MRI examination [3,6]. This study did not compare MRI examination with other imaging modalities. MRI examination is known to have a sensitivity and specificity of 100% in diagnosing MRKH syndrome. MRI imaging modalities are also known to have the same effectiveness when compared to a combination of clinical examinations and ultrasound examinations [7].

One of the characteristics of female patients with MRKH syndrome is the presence of ectopic ovaries with underdeveloped uterus. This is often mistaken to mean that patients with MRKH syndrome do not have a uterus at all [8]. In this patient the uterine organs developed abnormally which required a uterine transplant. In previous studies it can also be stated that it can be accompanied by agenesis of the vaginal organs as well [9].

MRKH syndrome is most frequently diagnosed in female adolescent patients with primary amenorrhea accompanied by the development of normal secondary sex characteristics in health facilities. Pelvic ultrasound is the main modality chosen to be performed in patients with these clinical symptoms. Furthermore, if it cannot be visualized properly, it will be done with an MRI examination [10]. Misdiagnosis is also common where doctors make further observations and do not provide early therapy so that there is a delayed diagnosis. Women with MRKH syndrome have never had menstruation or pregnancy. Even though ovarian cell donors and in vitro fertilization can be carried out, the patient may still feel depressed. In patients with vaginal agenesis, surgery is also needed so that they can have sexual intercourse with partners [11].

In the uterus of a normal person, an MRI examination can see 3 layers of the uterus, which consist of the myometrium, junctional zone, and endometrium [12]. The 3 layers of the uterus can vary in patients with MRKH syndrome depending on the severity of the disease, which ranges from no differentiation to low signal intensity eccentric nodule rings in some patients with MRKH syndrome. In some cases, the presence of intraluminal blood or adenomyosis on MRI suggests functioning endometrium in a rudimentary uterus. However, this does not affect fertility because fertility in patients with MRKH will also be disrupted [11,12].

In previous studies, there was a correlation between the ovary and the rudimentary uterus, where the uterus is always in a caudal position to the ovary. This feature is important to note for the diagnosis in patients who have a small uterus, especially in ectopic located ovaries [13].

About 41% of patients had one or more ectopic ovaries, and this finding was more common than in other studies that found 14% and 16% of the ovaries in patients with MRKH were ectopic [12,13]. When an ectopic ovary is found, ovum cannot be accessed transvaginal, but an abdominal approach is possible [14]. The vagina can be assessed accurately with MRI. Although this can be easily assessed by physical examination, it is not appropriate in young, sexually inactive female patients. Knowledge of vaginal length can be useful in counseling patients and their families regarding future treatment needs. Vaginal dilation procedures can also be performed in patients with MRKH syndrome, but MRI imaging cannot be used to evaluate the success of the procedure [15].

## Conclusion

This case report discusses a 25-year-old woman with primary amenorrhea with MRKH syndrome. MRI examination can be used to confirmed the diagnosis and is better in terms of sensitivity and specificity when compared to other modalities.

## Patient consent

Written informed consent for publication of this case report has been obtained from the patient.

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